MaterniT® GENOME

CLINICAL UPDATE

Case study 1: advanced maternal age

FEMALE

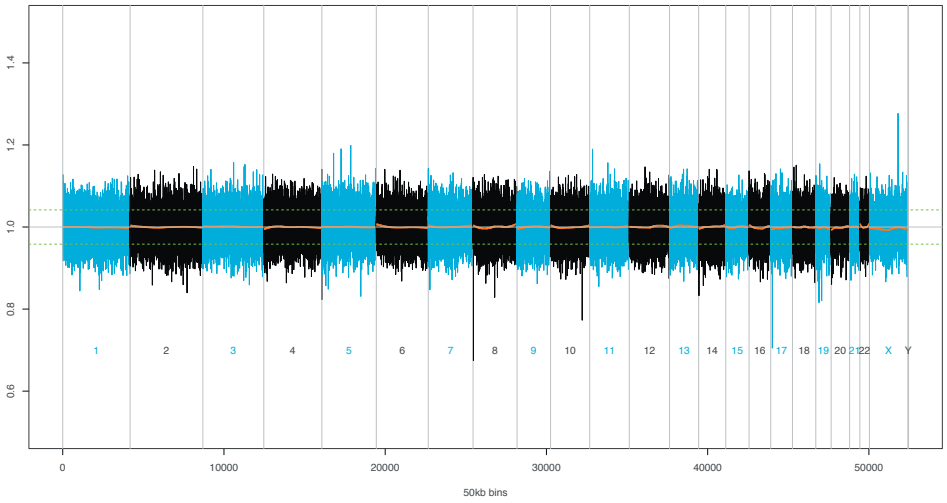
Advanced maternal age (AMA)

HISTORY OF MULTIPLE MISCARRIAGES

- First trimester ultrasound (12 weeks)
  - Nuchal translucency (NT) - Normal
- First trimester serum screen
  - Subsequently positive
  - Increased risk of trisomy 13 or trisomy 18

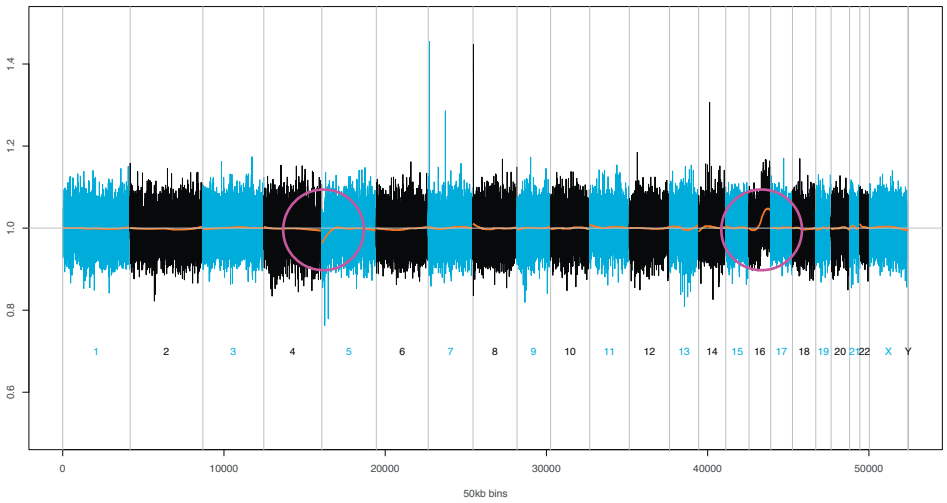
MATERNIT® GENOME ORDERED AT 12 WEEKS

Positive: 10.90 Mb loss at 5p15-ter and 30.85 Mb gain at 16q21-q24.3.



Normal 50 Kb trace (for comparison)

Each number below the trace represents a chromosome, from 1 to 22, X/Y. Note that the orange line stays relatively flat in a normal trace.



Positive MaterniT GENOME trace

Note the significant downward deviation on the orange line for chromosome 5p and upward deviation for chromosome 16q signifying a loss and gain of material, respectively.

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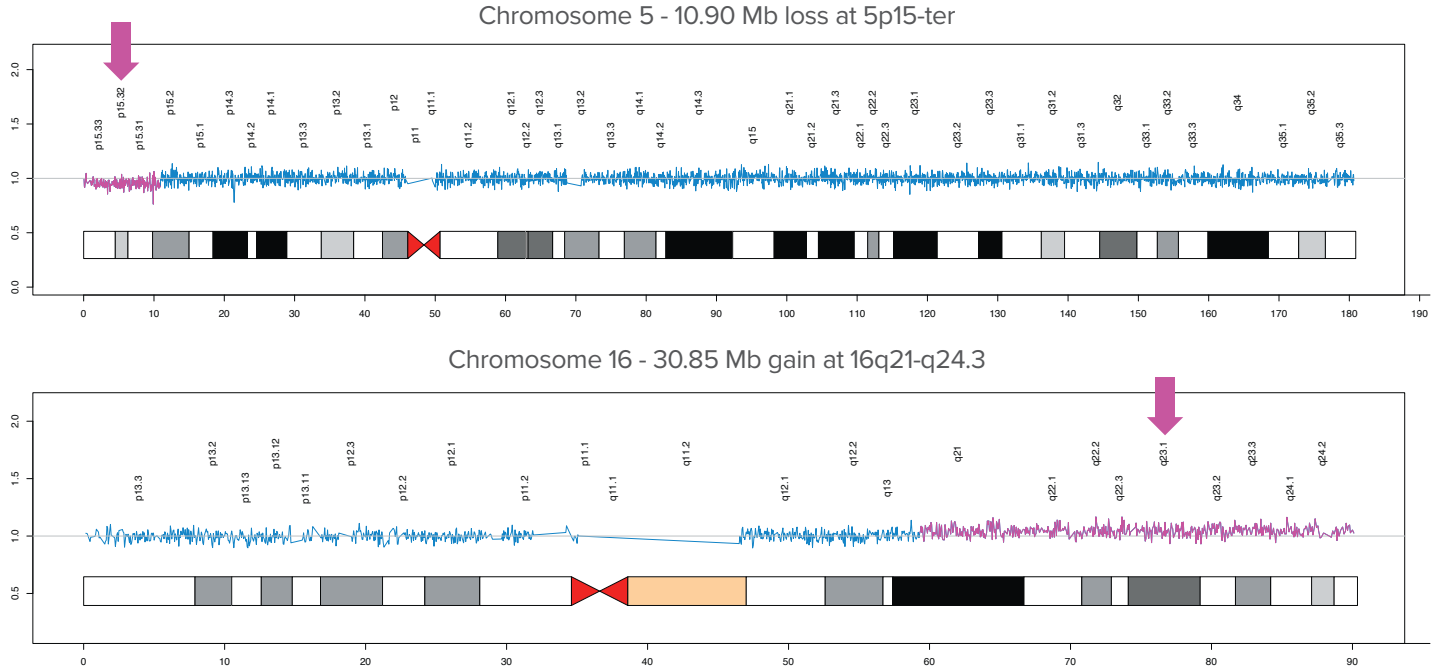
Key points

- Normal ultrasound > followed by abnormal serum screen > MateriT GENOME ordered at 12 weeks
- MateriT GENOME correctly identified chromosomal abnormalities different than those suggested by serum screen and in the presence of a normal ultrasound. Supported by confirmatory amniocentesis
- As illustrated by this case study, self-reported family history can be incomplete, and screening for only common aneuploidies (T13/18/21) with cfDNA may miss clinically relevant abnormalities on other chromosomes, potentially delivering false reassurance

MateriT GENOME detects up to 30% more chromosomal information than other NIPTs[1,2]; detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.



Ideograms from the MateriT GENOME lab report with closeup views of each impacted chromosomal trace provide a detailed view of the regions of interest. The resulting amplitude of the purple trace shows the deviations; in this case a loss on chromosome 5p and gain on chromosome 16q. Note the purple trace in relation to the blue trace.

Case study 1 summary

- Subsequent ultrasound at 16 weeks - Normal
- Amniocentesis at 16 weeks: Revealed abnormal result: 46,XX,der(5)**t(5;16)(p15.2;q21)**
- Unreported maternal translocation: **t(5;16)(p15.1;q21)** only disclosed by patient to Genetic Counselor after MateriT GENOME test results were issued
- Follow-up testing on POC (products of conception) confirmed amniocentesis results
- **MateriT GENOME test result confirmed by cytogenetics**

Results from case studies are not predictive of results in other cases. Results in other cases may vary.

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1. Ehrich M, Tynan J, Mazloom A, et al. Genome-wide cfDNA screening: clinical laboratory experience with the first 10,000 cases. *Genet Med.* 2017;19(12):1332-1337.
2. Internal data.